Infrarenal Aortic Rupture Secondary to Neurofibromatosis

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Neurofibromatosis is characterized by its cutaneous manifestations. It also is manifested by arterial lesions commonly found in arterioles and small arteries but rarely in large arteries. We present a patient with type I neurofibromatosis with spontaneous rupture of his abdominal aorta. He was found at the time of emergency laparotomy to have direct compression of his aorta by retroperitoneal neurofibromas and abnormal aortic structural wall integrity.

CASE REPORT

A 52-year-old white male with neurofibromatosis type I presented to the emergency department with a chief complaint of searing pain originating in his back and radiating to his abdomen in a belt-like distribution that originated 12 h earlier. He was reported to have collapsed earlier the same morning with possible loss of consciousness. After his collapse, the severity of this pain decreased but then worsened during the evening. He denied past history of cardiovascular disease, but used inhalers for chronic asthma.

On physical examination the patient was pale and clammy and in moderate distress with multiple pedunculated cutaneous lesions. Vital signs were blood pressure, 102/50 mmHg; pulse, 95 beats per minute; respiration, 16/min; and temperature, 98.8°F. His abdomen was flat, but there was definite left lower quadrant tenderness and guarding. His hemoglobin was 12.4 g/dL, hematocrit was 36.2%, and white blood count was 14.2 K/mm³. Coagulation studies were within normal limits. Abdominal radiographs revealed distended loops of small and large bowel. Spiral computed axial tomographic (CT) scan of the abdomen and pelvis was performed with oral contrast and showed a large retroperitoneal hemorrhage extending intraperitoneally around the liver and spleen. (Fig. 1). The hematoma contained both low- and high-density areas. The non-enhanced aorta appeared normal and there were numerous neurofibromas involving the lumbosacral spine (Fig. 2).

Three hours after admission, the patient reported a sudden onset of severely increased back and abdominal pain. His blood pressure decreased to 70/30 mmHg and his pulse increased to 130 bpm. He became unresponsive and developed agonal respirations, cyanosis, and abdominal distension. After intubation and rapid central administration of blood products and intravenous phenylephrine drip, he regained consciousness. A vascular surgical consultation was obtained and he was immediately taken to the operating room.

After mobilization of the bowel a large retroperitoneal hematoma was identified. The retroperitoneum was entered and a copious amount of blood was evacuated. A tear in his abdominal aorta distal to the origin of the renal arteries was discovered adjacent to extensive
neurofibromatosis. The neurofibroma tissue was debulked and bleeding was controlled with direct pressure. The aorta was cross-clamped but an attempt to primarily repair the tear was unsuccessful. Attempts to place a patch over the defect using a running Prolene suture failed because the aorta was extremely friable and the sutures tore through the aortic walls despite the use of Teflon pledgets. An intraoperative diagnosis of disseminated intravascular coagulopathy was made and blood products were administered to correct the coagulopathy. The abdomen was closed using silastic sheets since the bowel was grossly dilated. The patient expired an hour later.

**DISCUSSION**

Neurofibromatosis is an inherited hamartomatous disorder first described by Robert Smith in 1843 and later by Von Recklinghausen in 1882. The major characteristic component of this disorder is the neurofibroma, a tumor arising from fibroblasts of the neurilemmal sheath of peripheral nerves. There are at least eight variants of neurofibromatosis, but only two are well studied. Neurofibromatosis can be inherited in a simple autosomal dominant fashion, but at least 50% of cases are sporadic with a high mutation rate. Type 1 (NF-1), carried on chromosome 17, is the classic neurocutaneous disorder with an incidence of 1 in 3000 births. Type 2 (NF-2), carried on chromosome 22, is characterized by bilateral vestibular schwannomas and other central nervous system tumors and has an incidence of 1 in 40,000 births.

NF-1 occurs in 95% of patients with neurofibromatosis and there does not appear to be any correlation between particular genotypes and phenotypes. Although it is a congenital disease, most manifestations do not appear until childhood or early adult life. Manifestations may include learning difficulties, epilepsy, and mental retardation. Other aspects of the disorder include scoliosis, gastrointestinal neurofibromas, pheochromocytomas, and renal artery stenosis.

The cutaneous manifestations such as neurofibromas, café-au-lait spots, lentigines, and schwannomas are benign tumors originating in the neural sheath and commonly occur as solitary encapsulated subcutaneous tumors. The hallmark of NF-1 is the neurofibroma, an entity that is histologically distinct from the schwannoma. Although schwannomas can present with NF-1, its occurrence is rare. Our patient had the diagnosis of NF-1 confirmed on the basis of numerous cutaneous neurofibromas, café-au-lait spots, and an S1 schwannoma that was surgically removed 10 years prior to this admission.

Spontaneous hemorrhages, although rare, have been reported in neurofibromatosis in different body sites including the thoracic and abdominal cavities, retroperitoneum, and soft tissues of the trunk and extremities. Reubi developed one...